COL9A3 gene

collagen type IX alpha 3 chain

Normal Function

The *COL9A3* gene provides instructions for making part of a large molecule called type IX collagen. Collagens are a family of proteins that strengthen and support connective tissues, such as skin, bone, cartilage, tendons, and ligaments. In particular, type IX collagen is an important component of cartilage.

Type IX collagen is made up of three proteins that are produced from three distinct genes: one α1(IX) chain, which is produced from the *COL9A1* gene, one α2(IX) chain, which is produced from the *COL9A2* gene, and one α3(IX) chain, which is produced from the *COL9A3* gene. Type IX collagen is more flexible than other types of collagen molecules and is closely associated with type II collagen. Researchers believe that the flexible nature of type IX collagen allows it to act as a bridge that connects type II collagen with other cartilage components. Studies have shown that type IX collagen also interacts with the proteins produced from the *MATN3* and *COMP* genes.

Health Conditions Related to Genetic Changes

intervertebral disc disease

multiple epiphyseal dysplasia

At least three mutations in the *COL9A3* gene have been shown to cause dominant multiple epiphyseal dysplasia. All of these mutations disrupt how genetic information is spliced together to make the blueprint for producing the α3(IX) chain. These mutations, called splice-site mutations, change one DNA building block (nucleotide) near an area of the gene called exon 3. These mutations in the *COL9A3* gene result in the deletion of 12 protein building blocks (amino acids) from the α3(IX) chain. It is not known how mutations in *COL9A3* cause the signs and symptoms of dominant multiple epiphyseal dysplasia.

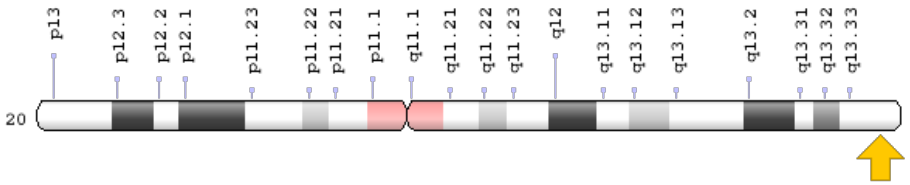
All identified mutations in type IX collagen delete a portion of the COL3 domain, which suggests that this region has an important function. Mutations may affect the ability of type IX collagen to fold correctly or interact with other cartilage components.

Stickler syndrome

Chromosomal Location

Cytogenetic Location: 20q13.33, which is the long (q) arm of chromosome 20 at position 13.33

Molecular Location: base pairs 62,817,062 to 62,841,159 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha 3 type IX collagen
- CO9A3_HUMAN
- collagen type IX alpha 3
- collagen, type IX, alpha 3
- DJ885L7.4.1
- EDM3
- FLJ90759
- IDD

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Collagens Are the Major Proteins of the Extracellular Matrix
  https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3551
- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix
  https://www.ncbi.nlm.nih.gov/books/NBK21582/

GeneReviews

- Multiple Epiphyseal Dysplasia, Dominant
  https://www.ncbi.nlm.nih.gov/books/NBK1123
- Stickler Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1302

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL9A3%5BTIAB%5D%29+OR+%28EDM3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D

OMIM

- COLLAGEN, TYPE IX, ALPHA-3
  http://omim.org/entry/120270

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=COL9A3%5Bgene%5D
- HGNC Gene Family: Collagen proteoglycans
  http://www.genenames.org/cgi-bin/genefamilies/set/575
- HGNC Gene Family: Collagens
  http://www.genenames.org/cgi-bin/genefamilies/set/490
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2219
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1299
- UniProt
  http://www.uniprot.org/uniprot/Q14050

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/24273071
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  https://www.ncbi.nlm.nih.gov/books/NBK1123
- Higashino K, Matsui Y, Yagi S, Takata Y, Goto T, Sakai T, Katoh S, Yasui N. The alpha2 type IX collagen tryptophan polymorphism is associated with the severity of disc degeneration in younger patients with herniated nucleus pulposus of the lumbar spine. Int Orthop. 2007 Feb;31(1):107-11. Epub 2006 Apr 4.
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